Important Information for Parents about the

Newborn Screening Test

California Department
of Health Services

Newborn Screening Program
Genetic Disease Branch
www.dhs.ca.gov/gdb

Publication Date: October 2005

The California Newborn Screening Test

Newborn screening can save your baby's life or prevent serious brain damage. Newborn screening can identify babies with certain diseases so that treatment can be started right away. Early identification and treatment can prevent mental retardation and/or life-threatening illness.

What Types Of Diseases Are Screened For In California?

To protect the health of all its newborns, California state law requires that your baby must have the Newborn Screening (NBS) Test before leaving the hospital. This test screens for specific diseases in the following groups:

Metabolic
chemical reactions in the body to create energy and build tissue

Endocrine
hormones that affect body functions

Hemoglobin
red blood cells that carry oxygen

In California there will be about 625 babies identified with one of these diseases each year. This means about 1 out of every 900 babies tested will have one of these diseases.

(See specific diseases under each category beginning on page 10.)

Make Sure Your Baby Is Tested

Babies with one of these diseases can look very healthy at birth and still have a serious disease. By the time symptoms appear, it may be too late to prevent serious damage to the baby. That is why your baby will be tested before leaving the hospital.

Can These Diseases Be Treated?

Yes. Effective treatment is available for most of the diseases for which we screen. Treatment may include special diets or drugs. Babies who receive early and ongoing treatment can grow up to enjoy long, productive lives. For some of the diseases found, there is no effective treatment.

What Is Screening?

Screening is the testing of a group of people to identify those who are at risk for having a specific disease even though they may seem healthy. Newborn screening identifies most, but not all, of the babies who have one or more of the many diseases screened for by the California Program. Not every baby with a positive screening test will have one of these diseases. Further testing and evaluation by the baby's health care provider or a specialist are needed to make the diagnosis.

How Is The Test Done?

A few drops of blood taken from the baby's heel are put on special filter paper. The blood is then sent to a state-approved lab for testing.

Is The Test Safe?

Yes, this is a simple and safe test. Over 12 million California newborns have had blood collected by heelstick without any harm to the newborn.

When Should The Test Be Done?

The newborn screening test should be done when the baby is at least 12 hours of age but before 6 days of age. Blood collected before 12 hours of age is **not** always reliable for some metabolic diseases. Another blood sample must be taken later to repeat the test. If you leave the hospital or birthing center with your baby before he/she is 12 hours old, you will have to return within the next few days for a second test.

Babies not born in the hospital must also have this test. It should be done by the time your baby is 6 days old. Call your midwife, the baby's doctor or your local health department to have your baby tested.

Can I Say No To The Test?

You can only say no for specific religious reasons. You must then sign a special form. It states that not having the test done can result in serious illness or permanent damage to your child. It also states that you accept responsibility should this occur.

Is The Test Accurate?

Yes. The blood is sent to a state-approved lab for testing. The state checks the work of the testing labs closely to make sure the results are reliable. It is rare that a baby with one of the more common diseases is not found through a positive newborn screening test. For a few rare diseases, the test may find only a small number of the babies affected.

How Can I Get The Results?

If the test is positive, you will be contacted within a few days after you leave the hospital. If the test is negative, it takes about two weeks for doctors to get a copy of the results. You can get your baby's test results from your doctor or clinic. If your doctor does not have the results, he/she can contact the Newborn Screening Program to request a copy.

If you move after the test is done, make sure the hospital and your baby's doctor or clinic have your new address and phone number in case they need to contact you about your baby's results.

What Do I Do If The Baby's Results Are Positive?

If the results are positive more tests will be needed. You should receive a phone call and/or letter about what to do next. After further testing, many babies who have a positive first test are found **not** to have a disease. However, you must have your baby re-tested because babies who do have one of these diseases benefit from early treatment.

Early Treatment Can Prevent Serious Problems

If these diseases are not found and treated soon after birth the baby can have serious health problems or even die. Early treatment can prevent many of these problems.

Metabolic Diseases

Metabolic diseases affect the body's ability to use certain parts of food for growth, energy, and repair. The parts include **amino acids** from proteins, **fatty acids** from fats and **organic acids** from proteins, fats, and sugars. To break down or convert these substances, certain proteins called enzymes must be present. When there aren't enough of the needed enzymes, some substances build up in large amounts and may be harmful to the body. Metabolic diseases have varying degrees of severity. If identified early, many of these conditions can be treated before they cause serious health problems. Treatment may include close monitoring of the person's health, medication, dietary supplements, and/or special diets.

These are some of the metabolic diseases screened for by the NBS Program:

- **Phenylketonuria (PKU)** – Babies with PKU have problems when they eat foods high in protein such as milk (including breast milk and formula), meat, eggs, and cheese. Without treatment babies with PKU develop mental retardation and/or have other health problems. A special diet can prevent these problems.

- **Galactosemia** – Babies with this disease cannot use some of the sugars in milk, formula and breast milk, and other foods. Without treatment babies with galactosemia can become very sick and die. A special infant formula and diet can help prevent these problems.

- **Maple Syrup Urine Disease (MSUD)** – Babies with MSUD have problems using fats and protein. Without treatment, MSUD can cause mental retardation or death. Treatment with a special diet can prevent these problems.

- **Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)** – Babies with MCADD are unable to convert fat into energy. Without treatment babies can have seizures, extreme sleepiness, coma, and even die. Treatment may include frequent feedings and a special diet.

- **Homocystinuria** – Babies with this disease have problems breaking down protein. Without treatment babies can have delayed development, eye problems, and/or other health problems. A special diet can help prevent these problems.

Endocrine Diseases

Babies with endocrine diseases make too much or too little of certain hormones. Hormones are produced by glands in the body and affect body functions.

- **Congenital Adrenal Hyperplasia (CAH)** – The adrenal glands of babies with this disease do not make enough of the key stress-fighting hormone cortisol. In about two-thirds of the cases, babies also do not produce enough of the salt-retaining hormone aldosterone. As a result, affected babies can develop dehydration, shock, and even death. Treatment with one or more oral medicines can help prevent these problems. Girls with this condition may have the additional problem of having masculine-looking external genitals, which can be corrected with surgery.

- **Primary Congenital Hypothyroidism** – Babies born with this disease do not have enough thyroid hormone. Without enough hormone, babies grow very slowly and have mental retardation. These problems can be prevented by giving the baby special thyroid medicine every day.

Hemoglobin Diseases

Hemoglobin is found in red blood cells. It gives blood its red color and carries oxygen to all parts of the body. Hemoglobin diseases often lead to anemia because they affect the type and amount of hemoglobin in the red blood cells. Treatment may include medication, folic acid and close monitoring of the child's health.

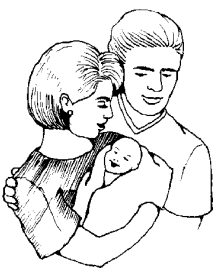
These are some of the hemoglobin diseases that are part of the newborn screening test:

- **Sickle Cell Anemia and other Sickle Cell Diseases** – These diseases affect the type of hemoglobin in the baby's red blood cells. Babies with sickle cell disease can get very sick and even die from common infections. Many of the infections can be prevented with daily antibiotics. Ongoing health care and close monitoring help children with hemoglobin diseases stay as healthy as possible.

- **Hemoglobin H Disease** – This disease affects the amount of hemoglobin in the baby's blood. There is less hemoglobin, which results in smaller red blood cells. This also causes the cells to break down faster than normal. Babies with this disease can have mild to severe anemia, as well as other health problems. Treatment can include blood transfusions, taking folic acid, and avoiding certain medications and household products.

Is Information About My Baby's Test Confidential?

Yes. For details of our privacy protection policies, read the notice included on pages 14-15 or on the website. There are serious penalties for any unauthorized release of private information collected during screening.



How Much Does The Test Cost?

The cost is subject to change. Please check with your doctor, the hospital, or the NBS website for the current cost of the test. Medi-Cal, health plans and most private insurance will pay for the test. The cost is included in the hospital bill. You will not receive a bill from the NBS Program. If you have problems with your insurance, contact 1-800-927-HELP (1-800-927-4357) or if you have a prepaid health plan, contact 1-888-HMO-2219 (1-888-466-2219).

California law prevents insurance companies from refusing to issue or canceling a policy, or charging a higher rate or premium based on a person's genetic characteristics, including being diagnosed with one of the diseases found by newborn screening. If you have any of these problems, call one of the numbers listed above. It is also illegal to refuse employment based on the results of a genetic test.

Does My Baby Need Any Other Blood Tests?

If there is something that you are worried about, or know of a disease that may run in the family, talk to your doctor about what other tests can be done. The Newborn Screening Program screens for the most common treatable diseases and includes almost all of the diseases a commercial newborn screening test would. The Program evaluates adding other diseases as new tests and treatments become available. However, the statewide program does not screen for every disease that might be found such as biotinidase deficiency and cystic fibrosis. In addition to screening, babies also need regular well-baby check-ups to see how the baby is growing, provide immunizations, and to look for these diseases as well as other health problems.

California Newborn Screening Program
Disorders That Can Be Detected As Of Mid-2005

Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Parents should remain watchful for any sign or symptoms of these disorders in their child and consult a physician.

I. Metabolic Disorders

A. Carbohydrate Disorders

- classical galactosemia

B. Amino Acid Disorders

- classical phenylketonuria (PKU)
- variant PKU
- guanosine triphosphate cyclohydrolase 1 (GTPCH deficiency) (biopterin deficiency)
- 6-pyruvoyl-tetrahydropterin synthase (PTPS deficiency) (biopterin deficiency)
- dihydropteridine reductase (DHPR deficiency) (biopterin deficiency)
- pterin-4α-carbinolamine dehydratase (PCD deficiency) (biopterin deficiency)
- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- citrullinemia, Type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia, Type II (citrin deficiency)
- gyrate atrophy of the choroid and retina
- homocitrullinuria, hyperornithinemia, hyperammonemia –HHH
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- methionine adenosyltransferase deficiency (MAT deficiency)
- maple syrup urine disease – (MSUD)
- tyrosinemia

C. Organic Acid Disorders

- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)

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- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
- 3-methylglutaconic aciduria (MGA), Type I (3-methylglutaconyl-CoA hydratase deficiency)
- beta-ketothiolase deficiency (BKT)
- ethylmalonic encephalopathy (EE)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- malonic aciduria
- methylmalonic acidemia, mut –
- methylmalonic acidemia, mut 0
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- multiple carboxylase deficiency (MCD)
- propionic acidemia (PA)

D. Fatty Acid Oxidation Disorders

- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/ glutaric acidemia type-2 (GA-2)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- trifunctional protein deficiency (TFP deficiency)
- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)

II. Endocrine Disorders

- primary congenital hypothyroidism
- variant hypothyroidism
- congenital adrenal hyperplasia-salt wasting (21-hydroxylase deficiency)
- congenital adrenal hyperplasia-simple virilizing (21-hydroxylase deficiency)

III. Hemoglobin Disorders

- sickle cell anemia (Hb S/S disease)
- sickle C disease (Hb S/C disease)
- sickle D disease (Hb S/D disease)
- sickle E disease (Hb S/E disease)
- Hb S/hereditary persistence of fetal hemoglobin (Hb S/HPFH)
- sickle cell disease variant (other sickle cell disease, Hb S/V)
- Hb S/Beta^0 thalassemia
- Hb S/Beta$^+$ thalassemia
- Hb C disease (Hb CC)
- Hb D disease (Hb DD)
- alpha thalassemia major
- Hb H disease

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- Hb H/Constant Spring disease
- beta thalassemia major
- Hb E/Beta^0 thalassemia
- Hb E/Beta$^+$ thalassemia
- Hb E/Delta Beta thalassemia
- Hb C/Beta^0 thalassemia
- Hb C/Beta$^+$ thalassemia
- Hb D/Beta^0 thalassemia
- Hb D/Beta$^+$ thalassemia
- Hb Variant/Beta^0 thalassemia
- Hb Variant/Beta$^+$ thalassemia
- other hemoglobinopathies (Hb variants)

For more information about the Newborn Screening Program and the most current list of diseases that can be detected through the Program, visit our website at

www.dhs.ca.gov/gdb

then click on Newborn Screening.

Questions for My Doctor

NOTICE OF INFORMATION AND PRIVACY PRACTICES
CA DEPARTMENT OF HEALTH SERVICES GENETIC DISEASE BRANCH
NEWBORN SCREENING PROGRAM (Effective Date April 14, 2003)
ABBREVIATED
(To request complete document, write to Department Contact below)

THIS NOTICE DESCRIBES HOW PERSONAL AND MEDICAL INFORMATION ABOUT YOU OR YOUR NEWBORN MAY BE USED AND DISCLOSED AND HOW YOU CAN GET ACCESS TO THIS INFORMATION. PLEASE REVIEW IT CAREFULLY.

Department's Legal Duty. Federal and State laws restrict the use, maintenance and disclosure of personal and medical information obtained by a State agency, and require certain notices to individuals whose information is maintained. In compliance with these laws, you and those providing information are notified of the following:

Department Authority and Purpose for the Newborn Screening Program. The Department of Health Services collects information related to newborn screening as permitted in Health and Safety Code Sections 124980, 125000, 125001, 125025, and 125030. This information is collected electronically and includes such things as your name, address, medical care given to you and your newborn. Testing is required by law (Health and Safety Code Section 125000) and regulations (17 CCR 6500 through 6510) and if the required information is not provided, death or permanent handicaps for affected newborns could result. If you have religious objections to testing, you may say "no" to testing in writing and sign a form advising you that your hospital, doctor and clinic staff are not responsible if your baby develops problems because those disorders were not identified and treated early.

Uses and Disclosure of Health Information. The Department of Health Services uses health information about you or your newborn for screening, to provide health care services, to obtain payment for screening, for administrative purposes, and to evaluate the quality of care that you or your newborn receives. Some of this information is retained for as long as 21 years. The information will not be sold.

The law also allows the Department to use or give out information we have about you or your newborn for the following reasons:

- For research studies unless you specifically request in writing that your information not be used.
- To organizations, which help us in our operations, such as collecting fees.

The information is otherwise confidential and will not be released without your written authorization. If you sign an authorization to disclose information you can later revoke that authorization to stop any future uses and disclosures by contacting the person listed below.

The Department may change its policies at any time subject to applicable laws and regulations. You may request a copy of our current policies or obtain more information about our privacy practices by contacting the person listed below or consulting our website at www.dhs.ca.gov/gdb. You may also request a paper copy of this notice.

Individual Rights and Access to Information. You have the right to look at or receive a copy (you will be charged) of your or your newborn's health information and receive a list of instances where we have disclosed health information about you or your newborn for reasons other than screening, payment or related administrative purposes. If you believe that information in your or your newborn's record is incorrect or missing, you have the right to request corrections. You have the right to make reasonable requests for us to contact you only in writing or at a different address, post office box, or telephone number.

You may request in writing that we restrict disclosure of your or your newborn's information for health care treatment, payment and administrative purposes. We are not required to agree to your request.

Complaints. If you believe that we have not protected your or your newborn's privacy or have violated any of your or your newborn's rights you may file a complaint by calling or writing: **Privacy Officer**, CA Department of Health Services, P.O. Box 997413, Sacramento, CA 95899-7413, 916-445-4646 or (877) 735-2929 TTY/TDD

You may also contact the Secretary of the Department of Health and Human Services, Office for Civil Rights at 50 United Nations Plaza, Room 322, San Francisco, CA. 94102, telephone (800) 368-1019. Or you may call the U.S. Office for Civil Rights at 866-OCR-PRIV (866-627-7748) or 866-788-4989 TTY.

The Department cannot take away your health care benefits or do anything to hurt you in any way if you choose to file a complaint or use any of the privacy rights in this notice.

Department Contact - The information on this form is maintained by the Department of Health Services, Genetic Disease Branch. The Chief of the Genetic Disease Branch is George Cunningham, M.D., 850 Marina Bay Parkway, F175, Mail Stop 8200, Richmond, California, 94804 (510-412-1502).

AMERICANS WITH DISABILITIES ACT
Notice and Information Access Statement

Policy of Nondiscrimination on the Basis of Disability
and Equal Employment Opportunity Statement

The Department of Health Services, State of California does not discriminate on the basis of disability in employment or in the admission and access to its programs or activities.

The Deputy Director, Office of Civil Rights, 1615 Capitol Avenue, Suite 73.720, Sacramento, CA 95814 has been designated to coordinate and carry out the agency's compliance with the nondiscrimination requirements of Title II of the Americans with Disabilities Act (ADA). Information concerning the provisions of the ADA, and the rights provided thereunder, are available from the ADA Coordinator.

The Genetic Disease Branch wants to provide quality services to the families of California and welcomes your comments and suggestions.

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Newborn Screening Program
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